

STEM CELL (BONE MARROW) TRANSPLANT

Information for people with Diamond Blackfan anemia, and their families.

What are stem cells?

All of the blood cells in the body start out as immature cells called blood-forming stem cells. Stem cells are able to grow into other blood cells that mature and function as needed in the body. Stem cells create the three main types of blood cells: red blood cells that carry oxygen throughout the body, white blood cells that fight infection, and platelets that help the blood to clot and prevent abnormal bleeding.

Where are stem cells located?

Stem cells are located in three places—bone marrow (the spongy center of the bone where blood cells are made), peripheral blood (found in blood vessels throughout the body), and cord blood (found in the umbilical cord and collected after a baby's birth). Stem cells for transplantation are obtained from any of these three places.

What is a stem cell transplant?

A stem cell transplant (also commonly referred to as a bone marrow transplant), takes healthy stem cells from a donor and gives them to the patient through a central line in a vein in the chest. The bag of stem cells usually looks similar to a bag of blood used for blood transfusion. This is because it contains red blood cells. The goal of a stem cell transplant is to replace unhealthy stem cells with new healthy ones. If all goes well, these healthy stem cells find their way to the bone marrow and begin to function and produce blood cells normally (called an engraft). It often takes several weeks for this to happen.

Why consider a stem cell transplant?

In Diamond Blackfan anemia (DBA), the bone marrow does not make enough red blood cells. Red blood cells carry oxygen to all of the organs in the body. When the number of red blood cells is low, the organs in the body do not get the oxygen they need to function normally. A stem cell transplant can help restore the marrow's ability to make red blood cells, and it is currently the only known cure for DBA. However, it is an expensive and potentially dangerous procedure that can lead to death or severe chronic illness in some patients. For this reason, stem cell transplant typically is not a "first line" treatment. Other treatments, such as steroid medicine (corticosteroid) therapy and blood transfusion therapy, tend to be used first, if possible. Stem cell transplants are used for some patients whose marrow no longer produces enough blood cells.

Techniques such as stem cell transplant continue to be refined and improved and may be considered in some cases. Before deciding to have a transplant, people with DBA should discuss the pros and cons of this procedure with their medical team.

Who can be a stem cell donor?

For a person to be a donor, the donated stem cells must closely match the patient's Human Leukocyte Antigen (HLA) type. HLA markers are special proteins found on most cells in the body. The immune system uses these proteins — or markers — to recognize which cells belong in the body and which do not. These markers are inherited from both parents. Special tests called HLA typing or HLA tissue typing determine whether the patient and the donor cells match. Close family members such as brothers and sisters (but rarely parents) are often used as donors because they are most likely to match the patient's tissue type. Each sibling who has the same parents has a 25 percent chance of matching the patient's tissue type. However, if a sibling also has one of the DBA genes, it will be passed to the recipient during the transplant. It is important to screen potential donors for DBA genes because there is a risk of transfer from a sibling who has the gene for DBA, but who has no symptoms.

For more information call:

1-877-DBA-NURSE





If there is not a brother or sister or other family member who is a match for the patient, the transplant center can check the National Marrow Donor Program (NMDP) registry for an unrelated matching donor. In some instances, unrelated donors may be adequately matched and able to donate. However, the rate of successful transplant from matched unrelated donors (MUDs) is lower. The best scenario is an identically matched, sibling who does not have DBA.

The National Marrow Donor Program (NMDP) is a database containing the tissue types of more than six million potential volunteer donors. Visit the program online to learn more: <http://www.marrow.org/index.html>.

What are the benefits of a stem cell transplant?

For DBA patients, a stem cell transplant is intended to restore the marrow's ability to make red blood cells. Once the body starts producing red blood cells, the patient may experience a decrease in signs and symptoms of anemia, such as tiredness and paleness. Often times, stem cell transplant may result in a cure of DBA and, when successful, may often extend a person's life and improve the quality of life they are able to enjoy. The person will no longer require long-term steroid medicine or blood transfusions. The person's blood type will actually change to that of the donor.

What are the risks of a stem cell transplant?

A stem cell transplant is a complex procedure with risks. Although some people with DBA experience few problems with transplant, others experience many problems and must endure frequent tests and hospitalizations. Before a stem cell transplant, the patient receives chemotherapy and occasionally radiation therapy to destroy their unhealthy stem cells. This is called a preparative regimen. Some side effects, such as nausea, vomiting, fatigue, loss of appetite, mouth sores, hair loss, and skin reactions may be due to the preparative regimen. It is important for the patient to maintain good nutrition throughout the transplant process. Several complications, some potentially fatal, can occur as a result of a stem cell transplant:

Infection

After the transplant, before the new marrow has started to grow, the number of white blood cells is low and the immune system (how the body fights infection and stays healthy) is very weak. During this time, the body is susceptible to infections, sometimes from the bacteria that live in the patient's own body. Therefore, infections that normally would not be harmful can be very serious, and patients can die of them. Bacterial, viral, and fungal infections are often seen following transplant.

Graft-versus-host disease (GVHD)

Graft-versus-host disease (GVHD) occurs when the new stem cells (from the donor) do not recognize the patient's cells and attacks them, leading to skin rashes, diarrhea, or liver abnormalities. GVHD can be acute or chronic and range in severity from mild to moderate to severe. Medicines are given to prevent GVHD. Mild and moderate GVHD can be treated successfully with drugs and does not increase the risk of the patient dying. The most severe degree of GVHD is less frequent, but very serious, and patients can die of this complication. A close match between the donor and recipient will reduce the risk for GVHD, thereby allowing a greater chance for the donor stem cells to produce normal blood cells without complications.

Transplant rejection (Graft Failure)

Graft failure happens when the patient's immune system attacks the new stem cells from the donor. This happens because the patient's immune system thinks the new cells are harmful and need to be destroyed. If the immune system wins the fight, the old stem cells will come back, along with the DBA.

Other risks

Some of the more common long-term risks of stem cell transplant include infertility (the inability to produce children) and cataracts (clouding of the lens of the eye, which can be fixed with surgery). Less common effects include long-term damage to organs such as the liver, kidneys, lungs, or heart, and the occurrence of cancers.

Will a stem cell transplant cure DBA?

Stem cell transplant is a potentially curative but dangerous procedure. It can lead to normal red blood cell



production, but it is risky. Matching bone marrow donors take time to find, and patients sometimes die of complications of the treatment. A stem cell transplant also can lead to severe chronic illness (such as GVHD) in some patients. Physical problems associated with DBA but not related to the bone marrow, such as a cleft palate or a heart defect, will not change. In addition, the person's genes will still have DBA, so there is still a 50 percent chance of passing the disorder to any future children, if fertility is retained.

Is follow-up care necessary? What does it involve?

Regular check-ups are needed to identify and take care of any problems that may arise after a patient has a stem cell transplant. Initially, follow-up care involves clinic visits once or twice a week with platelet or blood transfusions, as needed. Long-term follow-up is necessary to maintain a healthy lifestyle, ensure that the DBA continues to be in remission, and ensure that any late effects of the transplant or DBA are caught early. During long-term follow up, growth and development, immunizations, fertility, and mental and physical health are monitored.

Where can I learn more information about stem cell transplant?

- Blood and Marrow Transplant Information Network
<http://www.bmtinfonet.org/>
- Children's Hospital Boston
<http://www.childrenshospital.org/clinicalservices/Site1977/mainpageS1977P0.html>
- Children's Hospital & Research Center at Oakland
<http://hemonc.cho.org/BMTGeneral/overview-public.html>
<http://hemonc.cho.org/BMTGeneral/treatment.html>
- Cincinnati Children's Hospital Medical Center Bone Marrow Failure Clinic
<http://www.cincinnatichildrens.org/svc/alpha/b/blood/programs/bmf-clinic>
- Lucile Packard Children's Hospital at Stanford
<http://www.lpch.org/DiseaseHealthInfo/HealthLibrary/hematology/bonetran.html>
- Mayo Clinic
<http://www.mayoclinic.com/health/stem-cell-transplant/CA00067>
- Memorial Sloan-Kettering Cancer Center
<http://www.mskcc.org/mskcc/html/70018.cfm>
<http://www.mskcc.org/mskcc/html/9430.cfm>
- National Cancer Institute
http://www.cancer.gov/images/Documents/4d9c7199-895b-4e6f-a914-5c8226f2abeb/fs7_41.pdf
- University of Minnesota
<http://www.med.umn.edu/print/BMT/bmtfaq/faqgeneral.html>
<http://www.med.umn.edu/print/BMT/bmtfaq/faqcomplications.html>

Important things to remember

- Ask questions. Make a list ahead of time to discuss with the doctor at your next visit.
- Be your own advocate because many doctors are not familiar with DBA. Do your homework and be part of the decision-making process. Contact a DBA resource center if you have questions. Please see the Resources fact sheet for a list of DBA resource centers.
- There are risks and benefits to any treatment for DBA. The risks of a stem cell transplant are very serious and should be considered carefully.
- Ask for help. Support can come from friends, family members, doctors and nurses on your medical team, or a support group for people with DBA.
- Stay positive! Remember, you are not alone. There are many DBA specialists and support groups available to help you.